

Newborn Screening Expanded Test List

The MDCH Newborn Screening Laboratory has been conducting a pilot project to evaluate screening for additional disorders from tandem mass spectrometry since April 18, 2005. The new disorders are on a list commissioned by the Health Resources and Services Administration (HRSA) and developed by the American College of Medical Genetics in 2005. Their paper, entitled Newborn Screening: Toward a Uniform Screening Panel and System, can be found at <http://mchb.hrsa.gov/screening/>.

The list of disorders recommended for screening was the result of a comprehensive effort that reviewed 84 conditions. These disorders were ranked according to their clinical characteristics, the diagnosis, follow-up, treatment, management of the condition, and analytical characteristics of the test. They were assigned to one of three groups; a core panel (screened directly), secondary targets (available through multiplex testing), or inappropriate for screening.

The core panel, recommended by the March of Dimes and HRSA, consists of the 11 disorders Michigan previously reported, an additional 17 disorders detected with tandem mass spectrometry, cystic fibrosis, and newborn hearing screening. There are 25 secondary target disorders on the list and 21 of them were in the pilot evaluation. From April 18, 2005 to May 2006, a summary report was provided on the pilot conditions. Following a major upgrade to our Laboratory Information System, the laboratory formally began providing a comprehensive report of the all pilot disorders from tandem mass spectrometry in May 2006.

The large number of disorders (49) was condensed to six groups to fit on one report page. The patient numeric results and expected results are listed only when abnormal findings are observed; otherwise, patient results appear as "Within Normal Limits" for that analyze/disorder group. The disorder/analyte(s) groupings are amino acid disorders, fatty acid oxidation disorders, organic acid disorders, endocrine disorders, enzyme disorders, and hemoglobinopathies.

. You can review the complete list of disorders now being screened at http://www.michigan.gov/mdch/0,1607,7-132-2942_4911_4916-64851--,00.html.